

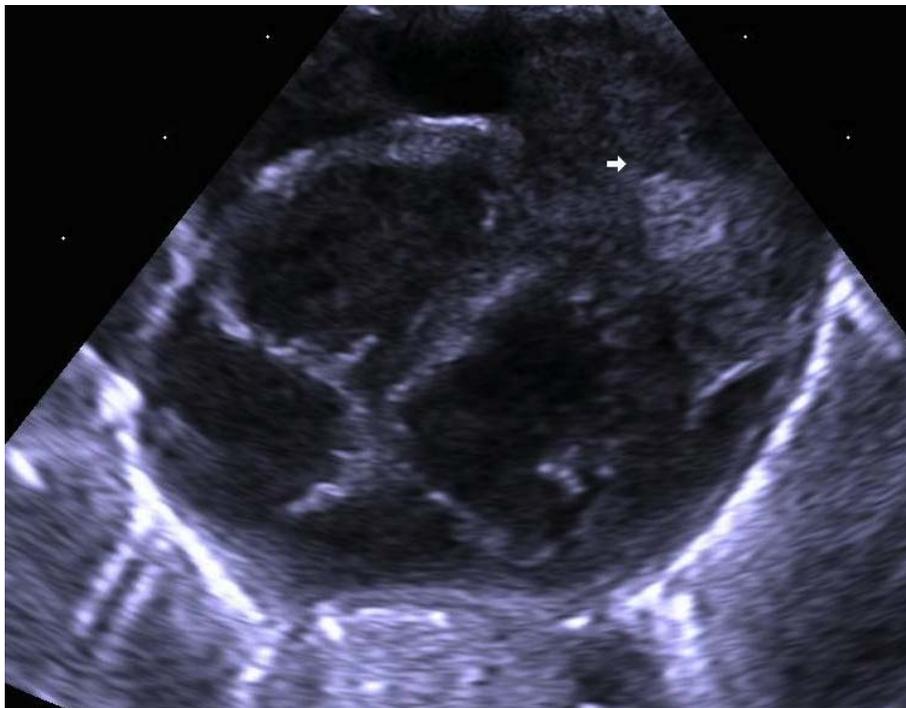
Gorlin syndrome: a familial diagnosis

*Recher M.(1), Rakza T.(1), Boute O.(2), Guillaume M.P.(1), Godart F.(1)
Paediatric Cardiology, Faculty of Medicine Lille 2, France(1), Genetics, Faculty of Medicine Lille 2,
France (2)*

Gorlin syndrome (or nevoid basal cell carcinoma syndrome) is characterized by a wide range of developmental abnormalities with prevalence from 1/57.000 to 1/256.000, a male-to-female ratio of 1:1. It is caused by mutations in the PTCH1 gene and is transmitted as an autosomal dominant trait with complete penetrance and variable expressivity. This case report describes a familial Gorlin syndrome discovered in utero by a cardiac tumour.

A cardiac tumour localized at the apex, septum and lateral wall of the left ventricular was discovered on antenatal echocardiography in a girl, associated with ventricular septal defect and a cerebral ventricular dilatation. A Gorlin syndrome was suspected but no genetic study was realized. In the familial history, she had a sister who presented a syndrome involving atrial septal defect, microcephaly, psychomotor retardation and epilepsy. She died accidentally at 4 years of age but no genetic study for Gorlin syndrome was realized. Our patient was born at 37 weeks without hemodynamic or respiratory disorders. The postnatal echocardiography confirmed the cardiac tumour at apex of the left ventricular and the muscular ventricular septal defect, associated with other anomaly (facial dysmorphism, talus valgus and moderate cerebral ventricular dilatation). A thoracic MRI showed the cardiac tumour in the apex, septum and the free wall of the left ventricular without hemodynamic obstruction. A biopsy was realized showing a fibroma on histology as usual in this syndrome. The genetic test was confirmed the Gorlin syndrome in this girl, and the same mutation was observed in her mother and her grandmother. The one year follow up showed no evolution of the tumour without hemodynamic disorder, but few briefs episodes of sinus tachycardia.

The combination of cardiac tumour and cerebral ventricular dilatation in utero should evoke the diagnosis of Gorlin Syndrome that could be confirmed by the appropriate genetic test.



Echocardiography of tumour